association of Ireland

# NEUROFIBROMATOSIS

***Information, help and support for those who need it***

association
of Ireland

*Our primary aim is to inform sufferers and their families of the disorder, supporting them and letting them know they are not alone with the condition.*

*If we can help you, someone you care about or you are looking for more information about NF then please do not hesitate to contact us.*

**Email:**
info@nfaireland.ie

**Helpline:**
085 702 0024

**Address:**
Neurofibromatosis Association of Ireland,
Carmichael Centre,
North Brunswick St,
Dublin 7

Reg Charity: No 6657
Company Reg No 299875

## WHAT IS NF

Neurofibromatosis (NF) is a genetic condition. This means it belongs to a group of health conditions that can be passed on in families from one generation to the next through the process of genetic inheritance.

NF is caused by a mutation in one of the genes. About half of the people who have NF have no family history. In other words, it has occurred "out of the blue" with no-one else in the family being affected. This is called a spontaneous gene mutation. The other half of people will have inherited NF from their mother or father.

NF occurs in all races. It affects both men (and boys) and women (and girls) equally. NF varies from one person to another even in the same family. Some people will be mildly affected with very few health problems. Others will have some serious health problems that mean that daily life is difficult and it restricts what they can do. With a condition that varies so much, it is important to learn some basic facts about NF, and to understand when you need to seek help from your doctor. It is important therefore that you are clear about the diagnosis that you have been given.

## COMMON SIGNS OF NF1

**GPs will refer patients to a geneticist or dermatologist to confirm the diagnosis of NF1. The condition is diagnosed by the presence of 2 or more of the following criteria, provided that no other disease accounts for the findings.**

**Café-au-lait patches, 6 or more** are one of the most common signs of NF1, flat pigmented spots on the skin. They tend to be a few shades darker than the usual colour of a persons skin. These spots are harmless and often help determine the diagnosis of NF1.

**Café-au-lait patches** (6 or more)

**A number of features commonly associated with NF1 are described below. An individual will not necessarily develop all these features.**

Freckling in specific body areas also occur in individuals with NF1. In those who do not have NF1, freckling usually occurs in areas exposed to sun; with NF1, freckling can be present in other areas, including the armpit (axillary freckling) and the groin (inguinal freckling). The freckles are often first noted around 3 or 4 years of age. Such freckling is not seen in every person with NF1, but it is considered strong evidence of the condition.

Lisch nodules are clumps of pigment in the coloured part of the eye (iris) that usually appear around puberty. They do not cause medical problems or affect vision. The presence of Lisch nodules can be helpful in confirming the diagnosis of NF1. Lisch nodules can be distinguised from iris freckles (commonly seen in people without NF1) by an ophthalmologist.

Neurofibromas, the most common tumours in NF1, are benign growths that typically develop on, or just

underneath, the surface of the skin; however, they may also occur in deeper areas of the body. Neurofibromas are composed of tissue from the nervous system (neuro) and fibrous tissue (fibroma). There are 2 major types of neurofibromas.

Dermal neurofibromas, also known as cutaneous neurfibromas, are small, nodule-like tumours on the surface of the skin. These may appear at any age, though are most likely to start developing in adolescence. Dermal neurofibroma rarely, if ever, become cancerous.

Plexiform neurofibromas grow diffusely or as nodules under the skin surface or deeper in the body. They may be present from birth, but not initially be noticeable. Plexiform neurofibromas can develop in any part of the body and tend to grow and intertwine with normal body tissues. They have approximately a 10% chance of becoming malignant. Sudden growth or pain in a plexifrom neurofibroma should be investigated by a physician.

The presence of multiple neurofibromas is an important diagnostic sign of NF1. (A single neurofibroma may occur in a person who does not have NF). The number of neurofibromas varies widely among affected individuals - from only a few to several hundred in very rare cases.

## WHAT TREATMENT IS AVAILABLE?

There is no cure for NF but most problems associated with the condition can be treated successfully. Due to the unpredictability of NF the assistance of one or more of the following specialists could be required: Paediatric, Dermatology, Endocrinology, Neurology, Ophthalmic Surgery, Orthopaedic Surgery, Oncology, Neurosurgery, & Plastic Surgery.

## INFORMATION & LITERATURE

The following brochures on NF can be obtained from the Neurofibromatosis Association, the Neurofibromatosis Clinics or through our Web Site.

- **Overview of Neurofibromatosis Type 1**
- **A Guide for Educators**
- **Learning and Cognitive Difficulties**
- **Neurofibromatosis Type 1 for Teens**
- **The Child with Neurofibromatosis Type 1**
- **Talking to Your Child**
- **Information, help and support for those who need it**
- **Clinical guidelines for the management of individuals with NF1**
- **Review Checklist for Adults & Children**

## NF2 BOOKLETS

- **Booklets for Families**
- **Booklets for Health Professionals**

## WHAT ARE THE TYPES OF NF?

NF manifests itself in many ways, there are 2 main Types, NF1 and NF2. NF1 is more common than NF2 and it can affect any part of the body. The Gene for NF1 is located on Chromosome 17.

**Problems to look out for in NF type 1:**

- **Optic Glioma**
- **Scoliosis (curvature of the spine)**
- **Learning & co-ordination problems**
- **Neuro-fibromas growth of tumours on nerve tissue**
- **High blood pressure**

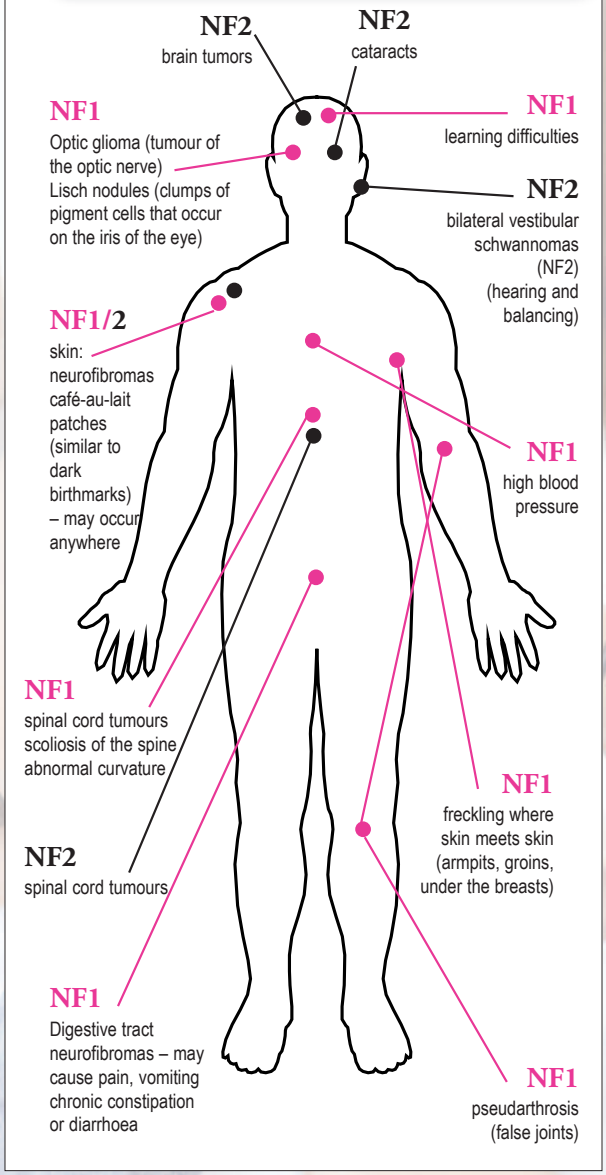
## NEUROFIBROMATOSIS TYPE 2

Another rarer type of Neurofibromatosis and distinct in its clinical feature is NF2. The gene for NF2 is located in chromosome 22. NF2 is generally a teenage and adult disease. NF2 Occurs in one in every 40,000 births.

**Common signs of NF2 include:**

- **Vestibular schwannoma (tumour on hearing nerve)**
- **Schwannoma (type of tumour of the substance that covers nerves fibres)**
- **Meningiomas (tumour of the covering of the brain)**
- **Cataract**

## How NF can affect the body



## NEUROFIBROMATOSIS CLINICS

**Both Adults and Children can be referred to:**
Department of Clinical Genetics
Children's Health Ireland at Crumlin, D12 N512
Telephone: + 353 1 409 6739

**Children under the age of 16 can be referred to:**
Tallaght NF Clinic
Children's Health Ireland at Tallaght University Hospital, D24 NR0A
Telephone: + 353 1 414 2000 / Bleep 7186

**Neurofibromatosis Type 2 Patients**
Neurosurgery and ENT at Beaumont Hospital, jointly manage the care of NF2 patients.

**Those attending an NF Clinic will need an appointment and a referral letter from their GP or Consultant.**

## MEDICAL EVALUATION & FOLLOW-UP

**Neurofibromatosis Type 1 (NF1)**
The initial examination is best carried out by a health professional who is knowledgeable about NF and its varied complications. In the case of GP's, they generally refer patients to a geneticist or consultant to confirm the diagnosis. Your consultant will advise on medical intervention and follow up care.

Checks in children with NF1 will be made for height, weight, head circumference, blood pressure, vision and hearing. Signs of learning disability, hyperactivity and evidence for scoliosis in addition to café-au-lait patches and neurofibromas will also be investigated.

As the condition is so variable in its presentation evaluation will include a detailed family history, ideally involving other family members to be examined to exclude NF.

Routine check-ups (6-12 months) for adults with NF1 generally include examination of the skin, blood pressure, and spine for scoliosis. Attention should also be given to any mass that is rapidly enlarging and causing new pain. (Ref, check list for adults & children).

**Neurofibromatosis Type 2 (NF2)**
Once a person has been diagnosed as having NF2 it is recommended that they have a full initial assessment, which includes a detailed neurological examination (testing balance and the strength and sensation in their bodies). This allows the person and the doctors to assess how NF2 is affecting them and to plan management.

**Having regular MRI scans and neurological examinations enables ongoing assessment of the growth of tumours and their affect on the person.**

## HOW YOU CAN HELP

- **BECOME A MEMBER**
- **JOIN THE COMMITTEE**
- **MAKE A DONATION**
- **FUNDRAISE FOR NF**

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